General

Guideline Title

Prenatal screening for and diagnosis of aneuploidy in twin pregnancies.

Bibliographic Source(s)

Audibert F, Gagnon A. Prenatal screening for and diagnosis of aneuploidy in twin pregnancies. J Obstet Gynaecol Can. 2011 Jul;33(7):754-67. [91 references] PubMed

Guideline Status

This is the current release of the guideline.

This guideline updates a previous version: Summers AM, Langlois S, Wyatt P, Wilson RD, Society of Obstetricians and Gynaecologists of Canada. Prenatal screening for fetal aneuploidy. J Obstet Gynaecol Can 2007 Feb;29(2):146-61.

Recommendations

Major Recommendations

The quality of evidence (I-III) and classification of recommendations (A-L) are defined at the end of the "Major Recommendations."

Prenatal Screening in Twins

Aneuploidy Risk Estimation in Relation to Maternal Age, Zygosity, and Chorionicity

Recommendations

1. All pregnant women in Canada, regardless of age, should be offered, through an informed counselling process, the option of a prenatal screening test for the most common clinically significant fetal aneuploidies. In addition, they should be offered a second trimester ultrasound for dating, assessment of fetal anatomy, and detection of multiples. (I-A)
2. Counselling must be non-directive and must respect a woman's right to accept or decline any or all of the testing or options offered at any point in the process. (III-A)
3. When non-invasive prenatal screening for aneuploidy is available, maternal age alone should not be an indication for invasive prenatal diagnosis in a twin pregnancy. (II-2A) If non-invasive prenatal screening is not available, invasive prenatal diagnosis in twins should be offered to women aged 35 and over. (II-2B)

Nuchal Translucency in Twins

Summary Statement

Fetal nuchal translucency combined with maternal age is an acceptable first trimester screening test for aneuploidies in twin pregnancies. (II-2)

Recommendations

4. Chorionicity has a major impact on the prenatal screening process and should be determined by ultrasound in the first trimester of all twin pregnancies. (II-2A)
5. When screening is done by nuchal translucency and maternal age, a pregnancy-specific risk should be calculated in monochorionic twins. In dichorionic twins, a fetus-specific risk should be calculated. (II-3C)

Nuchal Translucency Combined With Serum Markers

Summary Statement

First trimester serum screening combined with nuchal translucency may be considered in twin pregnancies. It provides some improvement over the performance of screening by nuchal translucency and maternal age by decreasing the false-positive rate. (II-3)

Integrated Screening

Summary Statement

Integrated screening with nuchal translucency plus first and second trimester serum screening is an option in twin pregnancies. Further prospective studies are required in this area, since it has not been validated in prospective studies in twins. (III)

Invasive Prenatal Diagnosis for Twin Pregnancies

Offering Invasive Testing in Multiple Pregnancies

Summary Statement

Non-directive counselling is essential when invasive testing is offered. (III)

Amniocentesis

Single or Double Sampling in Monochorionic Twins

Recommendation

6. During amniocentesis, both amniotic sacs should be sampled in monochorionic twin pregnancies, unless monochorionicity is confirmed before 14 weeks and the fetuses appear concordant for growth and anatomy. (II-2B)

Chorionic Villus Sampling

Chorionic Villus Sampling Error

Summary Statement

When chorionic villus sampling is performed in nonmonochorionic multiple pregnancies, a combination of transabdominal and transcervical approaches or a transabdominal only approach appears to provide the best results to minimize the likelihood of sampling errors. (II-2)

Management of Twins Discordant For Karyotypical Anomalies

Selective Reduction

Recommendation

7. Prior to invasive testing or in the context of twins discordant for an abnormality, selective reduction should be discussed and made available to those requesting the procedure after appropriate counselling. (III-B)

Technical Aspects

Recommendation

8. Monitoring for disseminated intravascular coagulopathy is not indicated in dichorionic twin pregnancies undergoing selective reduction. (II-

2B)

Quality of Evidence Assessment*

I: Evidence obtained from at least one properly randomized controlled trial

II-1: Evidence from well-designed controlled trials without randomization

II-2: Evidence from well–designed cohort (prospective or retrospective) or case–control studies, preferably from more than one centre or research group

II-3: Evidence obtained from comparisons between times or places with or without the intervention. Dramatic results in uncontrolled experiments (such as the results of treatment with penicillin in the 1940s) could also be included in this category.

III: Opinions of respected authorities, based on clinical experience, descriptive studies, or reports of expert committees

*Adapted from the Evaluation of Evidence criteria described in the Canadian Task Force on Preventive Health Care.

Classification of Recommendations†

A. There is good evidence to recommend the clinical preventive action.

B. There is fair evidence to recommend the clinical preventive action.

C. The existing evidence is conflicting and does not allow to make a recommendation for or against use of the clinical preventive action; however, other factors may influence decision-making.

D. There is fair evidence to recommend against the clinical preventive action.

E. There is good evidence to recommend against the clinical preventive action.

L. There is insufficient evidence (in quantity or quality) to make a recommendation; however, other factors may influence decision-making.

†Adapted from the Classification of Recommendations criteria described in the Canadian Task Force on Preventive Health Care.

Clinical Algorithm(s)

None provided

Scope

Disease/Condition(s)

- Fetal aneuploidy (e.g., Down syndrome and trisomy 18)
- Twin pregnancy

Guideline Category

Counseling

Diagnosis

Risk Assessment

Screening

Clinical Specialty

Medical Genetics

Obstetrics and Gynecology

Radiology

Intended Users

Advanced Practice Nurses

Physician Assistants

Physicians

Guideline Objective(s)

To provide a Canadian consensus document with recommendations on prenatal screening for and diagnosis of fetal aneuploidy (e.g., Down syndrome and trisomy 18) in twin pregnancies

Target Population

All pregnant women in Canada

Interventions and Practices Considered

1. Offering prenatal screening for aneuploidy to all pregnant women, regardless of age
    - Fetal nuchal translucency combined with maternal age for first trimester screening test
    - First trimester serum screening combined with nuchal translucency
    - Integrated screening with nuchal translucency plus first and second trimester serum screening
2. Use of nondirective counselling when offering testing options
3. Use of ultrasound in the first trimester to determine chorionicity
4. Calculation of pregnancy-specific risk in monochorionic twins and fetus-specific risk in dichorionic twins when screening is done by nuchal translucency and maternal age
5. Sampling from both amniotic sacs during amniocentesis in monochorionic twin pregnancies
6. Chorionic villus sampling in non-monochorionic multiple pregnancies (combination of transabdominal and transcervical approaches or a transabdominal only approach)
7. Counselling on selective reduction and ensuring its availability in those requesting the procedure
8. Monitoring for disseminated intravascular coagulopathy in dichorionic twin pregnancies undergoing selective reduction (considered but not recommended)

Major Outcomes Considered

- Incidence of use of different screening options in twin pregnancies
- Sensitivity, specificity, and positive predictive value of different screening options
- Pregnancy loss rate in chorionic villus sampling and amniocentesis

Methodology

Methods Used to Collect/Select the Evidence

Hand-searches of Published Literature (Primary Sources)

Searches of Electronic Databases

Searches of Unpublished Data

Description of Methods Used to Collect/Select the Evidence

PubMed and Cochrane Database were searched for relevant English and French language articles published between 1985 and 2010, using appropriate controlled vocabulary and key words (aneuploidy, Down syndrome, trisomy, prenatal screening, genetic health risk, genetic health surveillance, prenatal diagnosis, twin gestation). Results were restricted to systematic reviews, randomized controlled trials, and relevant observational studies. Searches were updated on a regular basis and incorporated in the guideline to August 2010. Grey (unpublished) literature was identified through searching the websites of health technology assessment and health technology assessment-related agencies, clinical practice guideline collections, clinical trial registries, and national and international medical specialty societies. The previous Society of Obstetricians and Gynaecologists of Canada guidelines regarding prenatal screening were also reviewed in developing this clinical practice guideline.

Number of Source Documents

Not stated

Methods Used to Assess the Quality and Strength of the Evidence

Weighting According to a Rating Scheme (Scheme Given)

Rating Scheme for the Strength of the Evidence

Quality of Evidence Assessment*

I: Evidence obtained from at least one properly randomized controlled trial

II-1: Evidence from well-designed controlled trials without randomization

II-2: Evidence from well–designed cohort (prospective or retrospective) or case–control studies, preferably from more than one centre or research group

II-3: Evidence obtained from comparisons between times or places with or without the intervention. Dramatic results in uncontrolled experiments (such as the results of treatment with penicillin in the 1940s) could also be included in this category

III: Opinions of respected authorities, based on clinical experience, descriptive studies, or reports of expert committees

*Adapted from the Evaluation of Evidence criteria described in the Canadian Task Force on Preventive Health Care.

Methods Used to Analyze the Evidence

Systematic Review with Evidence Tables

Description of the Methods Used to Analyze the Evidence

The quality of evidence was rated using the criteria described in the Report of the Canadian Task Force on Preventive Health Care (see the "Rating Scheme for the Strength of the Evidence" field).

Methods Used to Formulate the Recommendations

Expert Consensus

Description of Methods Used to Formulate the Recommendations

Not stated

Rating Scheme for the Strength of the Recommendations

Classification of Recommendations†

A. There is good evidence to recommend the clinical preventive action.

B. There is fair evidence to recommend the clinical preventive action.

C. The existing evidence is conflicting and does not allow to make a recommendation for or against use of the clinical preventive action; however, other factors may influence decision-making.

D. There is fair evidence to recommend against the clinical preventive action.

E. There is good evidence to recommend against the clinical preventive action.

L. There is insufficient evidence (in quantity or quality) to make a recommendation; however, other factors may influence decision-making.

†Adapted from the Classification of Recommendations criteria described in the Canadian Task Force on Preventive Health Care.

Cost Analysis

A formal cost analysis was not performed and published cost analyses were not reviewed.

Method of Guideline Validation

Internal Peer Review

Description of Method of Guideline Validation

This clinical practice guideline has been prepared by the Genetics Committee of the Society of Obstetricians and Gynaecologists of Canada (SOGC) and the Prenatal Diagnosis Committee of the Canadian College of Medical Geneticists (CCMG) and approved by the Executive and Council of the Society of Obstetricians and Gynaecologists of Canada and the Board of Directors of the Canadian College of Medical Geneticists.

Evidence Supporting the Recommendations

Type of Evidence Supporting the Recommendations

The type of supporting evidence is identified and graded for each recommendation (see the "Major Recommendations" field).

Benefits/Harms of Implementing the Guideline Recommendations

Potential Benefits

- Clinicians who are better informed about the accuracy of different screening options in twin pregnancies and about techniques of invasive prenatal diagnosis in twins
- Creation of specific guidelines for prenatal screening and diagnosis in twins
- Use of most accurate testing methods results in reduction of false-positive rate and improved detection rate of multiples and the most

common clinically significant fetal aneuploidies
- Improved counselling of women about prenatal screening and the most common clinically significant fetal aneuploidies

Potential Harms

- Risk of a false-positive result for prenatal screening tests for the most common clinically significant fetal aneuploidies
- The overall pregnancy loss rate associated with chorionic villus sampling (CVS) and amniocentesis in twin pregnancies appear to be similar, although the attributable loss rate has not been clearly defined for CVS.
- There is a risk of sampling error with both CVS and amniocentesis.

Contraindications

Contraindications

Methylene blue was first used as a dye in amniocentesis procedures, but its association with small bowel atresia and fetal death makes its use contraindicated.

Qualifying Statements

Qualifying Statements

This document reflects emerging clinical and scientific advances on the date issued and is subject to change. The information should not be construed as dictating an exclusive course of treatment or procedure to be followed. Local institutions can dictate amendments to these opinions. They should be well documented if modified at the local level. None of these contents may be reproduced in any form without prior written permission of the Society of Obstetricians and Gynaecologists of Canada (SOGC).

Implementation of the Guideline

Description of Implementation Strategy

An implementation strategy was not provided.

Implementation Tools

Foreign Language Translations

For information about availability, see the *Availability of Companion Documents* and *Patient Resources* fields below.

Institute of Medicine (IOM) National Healthcare Quality Report Categories

IOM Care Need

Staying Healthy

IOM Domain

Effectiveness

Patient-centeredness

Safety

Identifying Information and Availability

Bibliographic Source(s)

Audibert F, Gagnon A. Prenatal screening for and diagnosis of aneuploidy in twin pregnancies. J Obstet Gynaecol Can. 2011 Jul;33(7):754-67. [91 references] PubMed

Adaptation

Not applicable: The guideline was not adapted from another source.

Date Released

2007 Feb (revised 2011 Jul)

Guideline Developer(s)

Canadian College of Medical Geneticists - Professional Association

Society of Obstetricians and Gynaecologists of Canada - Medical Specialty Society

Source(s) of Funding

Society of Obstetricians and Gynaecologists of Canada

Guideline Committee

Society of Obstetricians and Gynaecologists of Canada Genetics Committee

Canadian College of Medical Geneticists Prenatal Diagnosis Committee

Composition of Group That Authored the Guideline

Principal Authors: François Audibert, MD, Montreal QC; Alain Gagnon, MD, Vancouver BC

Society of Obstetricians and Gynaecologists of Canada (SOGC) Genetics Committee Members: R. Douglas Wilson, MD (*Chair*), Calgary AB; François Audibert, MD, Montreal QC; Claire Blight, RN, Halifax NS; Jo-Ann Brock, MD, Halifax NS; Lola Cartier, MSc, CCGC, Montreal QC; Valerie A. Désilets, MD, Montreal QC; Alain Gagnon, MD, Vancouver BC; Jo-Ann Johnson, MD, Calgary AB; Sylvie Langlois, MD, Vancouver BC; Lynn Murphy-Kaulbeck, MD, Moncton NB; Nanette Okun, MD, Toronto ON; Melanie Pastuck, RN, Calgary AB; Vyta Senikas, MD, Ottawa ON

Canadian College of Medical Geneticists (CCMG) Committee on Prenatal Diagnosis Members: Sylvie Langlois, MD (*Chair*), Vancouver

BC; David Chitayat, MD, Toronto ON; Valerie A. Désilets, MD, Montreal QC; Michael T. Geraghty, MD, Ottawa ON; Janet Marcadier, MSc, Ottawa ON; Tanya N. Nelson, PhD, Vancouver BC; David Skidmore, MD, Halifax NS; Vicky Siu, MD, London ON; Frederique Tihy, PhD, Montreal QC

Financial Disclosures/Conflicts of Interest

Disclosure statements have been received from all members of the committees.

Guideline Status

This is the current release of the guideline.

This guideline updates a previous version: Summers AM, Langlois S, Wyatt P, Wilson RD, Society of Obstetricians and Gynaecologists of Canada. Prenatal screening for fetal aneuploidy. J Obstet Gynaecol Can 2007 Feb;29(2):146-61.

Guideline Availability

Electronic copies: Available in Portable Document Format (PDF) from the Society of Obstetricians and Gynaecologists of Canada (SOGC) Web site. Also available in French from the SOGC Web site.

Print copies: Available from the Society of Obstetricians and Gynaecologists of Canada, La société des obstétriciens et gynécologues du Canada (SOGC) 780 promenade Echo Drive Ottawa, ON K1S 5R7 (Canada); Phone: 1-800-561-2416.

Availability of Companion Documents

None available

Patient Resources

None available

NGC Status

This NGC summary was completed by ECRI Institute on February 5, 2009. The information was verified by the guideline developer on March 4, 2009. This NGC summary was updated by ECRI Institute on October 19, 2011. The updated information was verified by the guideline developer on November 14, 2011.

Copyright Statement

Disclaimer

NGC Disclaimer

Guidelines represented on the NGC Web site are submitted by guideline developers, and are screened solely to determine that they meet the NGC Inclusion Criteria which may be found at http://www.guideline.gov/about/inclusion-criteria.aspx.

NGC, AHRQ, and its contractor ECRI Institute make no warranties concerning the content or clinical efficacy or effectiveness of the clinical practice guidelines and related materials represented on this site. Moreover, the views and opinions of developers or authors of guidelines represented on this site do not necessarily state or reflect those of NGC, AHRQ, or its contractor ECRI Institute, and inclusion or hosting of guidelines in NGC may not be used for advertising or commercial endorsement purposes.

Readers with questions regarding guideline content are directed to contact the guideline developer.